GENDIA

Carrier Screening Test for Inherited Disorders (STID)

DESCRIPTION : STID (Screening Test for Inherited Disorders) is a test to screen for carriership of > 500 common and severe genetic disorders with recessive inheritance.
The STID is also referred to as the "carrier test".

WHY STID : STID screens healthy couples seeking children for carriership of > 500 common recessive disorders, including cystic fibrosis (CF), spinal muscle atrophy (SMA), sickle cell, thalassemia, mental retardation, blindness, deafness and many metabolic abnormalities.
The overall frequency of such recessive disorders is 1 %, which is higher than the frequency of Down syndrome.

If both partners carry a mutation in the same disorder gene, they have a 25 % risk that their offspring will be affected by a recessive disorder, and prenatal diagnosis with chorionic biopsy (CVS) or amniocentesis (AC) can be offered.

SAMPLE : 5 ml EDTA blood, or 5 ug DNA, or saliva in a special saliva kit (available at GENDIA), has to be sent at room temperature to GENDIA's lab in Antwerp, Belgium.

METHODS : Samples are analyzed by sequencing of 436 disease genes implicated in more than 500 common severe genetic disorders with recessive inheritance.

The test is carried out under CLIA and CAP accreditation.

TURNAROUND TIME : STID takes 1 month from arrival of the sample in the lab.

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INDICATIONS : STID can be performed in all pregnancies without specific indication, as recessive disorders occur in the offspring of a healthy couple that is unaware they carry a mutation in the same gene.
About 4% of the couples that are screened with the STID are both carriers of a pathogenic variant in the same gene. Because these couples have a 25% risk of having a child with this hereditary disease, they are offered a prenatal examination by chorionic villus sampling (chorion biopsy - CVS) or amniocentesis (amniocentesis - AC).

LIMITATIONS : Samples are only analyzed for variants in 436 disease genes implicated in > 500 common and severe genetic disorders with recessive inheritance.
Some variants in these genes or variants in other genes are not detected. Therefore, a normal STID test does not exclude carriership for a genetic disorder.

RESULTS : STID results will be sent to the patient and/or physician who ordered the test. Genetic counseling with explanation of the test results and recommended follow-up steps, if necessary, can be provided by genetic counselors from GENDIA.

PRICE : 390 Euro per person.

MORE INFO : See our website www.STID-GENDIA.net

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